DRAFT

Virginia Genetics Advisory Committee
Tuesday, October 16, 2007
10:00 a.m. – 12:00 Noon

Division of Consolidated Laboratory Services
Training Room T-23
600 North 5th Street
Richmond, VA 23219
Telephone (804) 648-4480

MINUTES

Present: VaGAC Members (check=present)

- ☑ Willie Andrews, DCLS
- ☑ Joann N. Bodurtha, VCU (VaGAC Chair)
- ☐ Joanne Boise, VDH (Ad Hoc Member)
- ☐ Nancy Bullock, VDH
- ☐ Tia Campbell DOE
- ☑ Lillie Chandler
- ☐ Joan Corder-Mabe, VDH (Ad Hoc Member)
- ☐ Mary Ann Discenza, DMHMRSAS
- ☑ Laura Duncan, VCU
- ☑ Nancy Ford, VDH (VaGAC Staff)
- ☑ Audrey Greene, VDH
- ☐ Anil R. Kumar, VCU
- ☐ Sara Long, MOD
- ☐ Walter Nance, VEHDIP Adv Cmt
- ☑ Arti Pandya, VCU
- ☐ James Pearson, DCLS
- ☑ Ginny Proud, CHKD
- ☑ Jene O. Radcliffe-Shipman, VDH
- ☑ Rafael Randolph, VDH
- ☐ Ramona Schaeffer, VDH
- ☑ Charlie Stevenson, DCLS
- ☐ David Suttle, VDH
- ☐ Sharon Williams, VDH
- ☑ Bill Wilson, UVA
- ☐ Melody Persinger Yeargin, CHKD

Present: Others (VaGAC Subcommittee members, guests)

- ☑ Stephen Braddock, UVA
- ☑ Bonny Bukaveckas, VCU
- ☑ Eileen Coffman, CHKD
- ☑ Sarah H. Elsea, VCU
- ☑ Mary Claire Ikenberry, VDOE (for T. Campbell)
- ☑ Rees Lee, NMCP
- ☑ Phil Poston, DCLS

Recorders: Rafael Randolph, Nancy Ford

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1. **Welcome: J. Bodurtha**
   a. Introductions: Completed.
   b. Minutes of 4/17/07 Meeting: Approved as written.
   c. Agenda: Reviewed, no new items added.
   d. Membership (revised 10/16/2007): Circulated for corrections.

2. **Cystic Fibrosis (CF): Rees L. Lee, M.D., Director, Navel Medical Center Portsmouth, CF Foundation Accredited Care Center**
   a. *Five CF Centers in Virginia:* (1) EVMS, Norfolk, (2) VCU Medical Center, Richmond, (3) Naval Medical Center Children and Adult Program, Portsmouth, (4) Pediatric Lung Center, Fairfax Neonatal Associates, and (5) UVA Children's Medical Center, Charlottesville.
   b. *CF Overview:* CF is an autosomal recessive genetic disorder, which means both copies of the gene in each cell have mutations. This disorder causes progressive damage to the respiratory system and chronic digestive system problems. CF is the most common genetic condition in

Caucasians. In the U.S., CF occurs in about 1 in 3,200 Caucasian newborns. CF is less common in other ethnic groups, affecting 1 in 15,000 African Americans and 1 in 31,000 Asian Americans. The median age of survival for people with CF is increasing due to early diagnosis and aggressive treatment.

c. *Benefits:* The following benefits are associated with Newborn Screening (NBS) for CF: improved growth, reduced hospitalizations, and improved survival. However, NBS does not pick up all CF; therefore, it is recommended that health care providers make the CF carrier screening test available to all couples. There is concern that NBS might negate diagnosis of CF not identified via NBS.

d. *Resource:* "Guidelines for Implementation of Cystic Fibrosis Newborn Screening Programs: Cystic Fibrosis Foundation Workshop Report," PEDIATRICS Vol. 119 No. 2 February 2007, pp. e495-e518 (doi:10.1542/peds.2006-1993). Published online February 1, 2007, http://pediatrics.aappublications.org/cgi/content/full/119/2/e495. Report outlines the steps necessary for success based on the experience of existing programs.

e. *Challenge:* Getting data from the five regional CF centers.

3. **VaGAC Infrastructure: J. Bodurtha**

a. Membership:
- *Newborn Screening Subcommittee:* Broadening membership beyond metabolic disorders experts and stakeholders (see Subcommittee Report).
- *Reminder:* All Subcommittee chairs need to send updated membership to N. Ford.
- *Nonattendees:* Discussed need to contact those who have not been attending to assess their interest in remaining on VaGAC.
- *Plan:* ☐ J. Bodurtha will contact nonattendees to address their remaining on VaGAC or other options for making contributions to VaGAC.

b. Meeting Setting:
- *Options:* Discussed advantages and disadvantages of various meeting options, such as face-to-face, online, and videoconferencing meetings.
- *Plan:* ☐ N. Ford will explore helpfulness of polycom options to promote attendance..

c. Meeting Time:
- *Schedule:* Discussed advantages and disadvantages of having meetings during the following times: (1) 10 a.m. – 12 p.m., (2) 10 a.m. – 2 p.m. including lunch, or (3) 10 a.m. – 1 p.m. with Subcommittee meetings following.
- *Decision:* Continue VaGAC current meeting time (10 a.m. – 12 p.m.) and continue option for Subcommittees to meet after VaGAC meeting with each chair making arrangements for lunch, location, etc.

d. Communication Access Realtime Translation (CART): Services will be made available for members and interested parties attending VaGAC meetings upon request to N. Ford.

4. **Subcommittees and Work Groups Reports**

a. **Newborn Screening Subcommittee: W. Andrews**
- *Meetings and Membership:* Quarterly meetings have been scheduled. These will be a combination of face-to-face and audio-conference call meetings. New members were added: Dr. Rees Lee (cystic fibrosis); Dr. Owen, CHKD (hemoglobinapathy disorders); and Dr. Cynthia Epstein, EVMS (cystic fibrosis). W. Andrews is waiting for responses from endocrinology consultants.
- *Public Health Laboratory Interoperability Project (PHLIP):* In May 2007, the Subcommittee recommended that DCLS participate in this international data collection project, which is a HRSA-funded grant conducted by Mayo Clinic for the purpose of evaluating newborn dried-blood-spot screening cut-off values for 42 disorders. Currently, 38

states are participating. P. Poston has started to submit Virginia data, which include only confirmed cases for those disorder detected by MS/MS technology. All data are anonymous.

- *Case Reviews:* The Subcommittee is considering reviewing cases quarterly to identify issues and prevent missed diagnoses.
- *Clinical and Laboratory Standards Institute (CLSI):* B. Wilson serves on CLSI's Subcommittee on Newborn Screening Guidelines for Premature and/or Sick Newborns. This is an international endeavor and a very complicated undertaking. According to CLSI—which is a global, nonprofit, standards-developing organization that promotes the development and use of voluntary consensus standards and guidelines within the healthcare community—this guideline will address best practice for NICU personnel, as well as primary health care providers, laboratory and follow-up personnel to provide all NICU infants with valid NBS within a reasonable amount of time. The primary goals for this project are to (1) increase consistency among neonatal intensive care practices for newborn screening specimen collection, (2) decrease the number of premature or sick newborns requiring multiple repeat specimen collection, (3) increase consistency among newborn screening follow-up systems for assuring rapid complete follow up for all NICU patients, (4) minimize the risk of a missed diagnosis for screened conditions, and (5) promote the need for further research on cause and effect of NICU treatments on newborn screening results for which these guidelines could not specify recommendations. Source: http://enews.nccls.org/clsi/issues/2007-04-01/7.html.

b. **Genetics and Public Health Subcommittee: J. Bodurtha**
   - *State Genetics Education Plan Work Group:* Governor Kaine has declared the month of November as Family Health History Month in Virginia. The VDH Family Health History from, which was developed as part of the Virginia Congenital Anomalies Tracking and Prevention Improvement Project (VaCATPIP), has been approved by VDH management for publication and distribution.
   - *State Genetics Plan:* The Virginia Genetics Program, with consultation provided by J. Bodurtha, is drafting a state genetics plan for public health. It is based on the core functions of public health (assessment, policy development, and assurance) and the ten essential public health services. A section on proposed future initiatives will serve as a starting point for further discussion with stakeholders, including VaGAC, on how best to meet identified needs related to furthering the mission of the Virginia Genetics Program.
   - *Genetics Needs Assessment*: The draft report "Genetics Needs Assessment: A Study of Genetic Services in Virginia," which summarizes results of a conducted by the VDH Division of Child and Adolescent Health and Division of Women's and Infants' Health, was completed April 2007 and is being finalized by VDH management (completion date to be determined). After finalized, the document will be shared with VaGAC.

c. **Community Partnerships Subcommittee, Parent Involvement Work Group: J. Monaco**
   - *Chair/Staff:* Allison Schreiber has resigned; the to-be-hire genetic counselor will be the new chair/staff.
   - *Meetings:* Work Group will not have face-to-face meetings; instead, work group will meet via conference calls as needed, which has worked well in the past.

d. **VDH Contractors Subcommittee: S. Williams**
   - *No report.*

5. **Updates: Agencies, Organizations, Grants**

a. **VDH: PSGS Grants VaCATPIP II, VaCHISIP, EHDI: N. Ford**
   - *Virginia Congenital Anomaly Tracking and Prevention Improvement Grant (VaCATPIP II):* The year-4 (budget period 3/1/2008 – 2/28/2009) continuation application for this five-year CDC-funded birth defects surveillance and evaluation grant, which supports VaGAC, is due November 15, 2007.

- *Virginia Child Health Information System Integration Project (VaCHISIP)*: The year-3 (budget period 7/1/2007 – 6/30/2008) continuation application for this three-year CDC-funded early hearing detection and intervention grant, which funds the redesign of the Virginia Infant Screening and Infant Tracking System (VISITS II), was submitted and awarded.
- *Universal Newborn Hearing Screening Grant*: The year-3 (budget period 9/1/2007 – 8/31/2008) continuation application for this three-year HRSA-funded grant, which funds the Virginia Guide By Your Side Program (GBYS) and Virginia Hearing Aid Loan Bank, was submitted and awarded.
- *Preventive Health and Health Services (PHHS) Block Grant:* A proposal was submitted to use PHHS funds, which are managed by VDH Office of Family Health Services, to support the Guide By Your Side program.
- *PSGS Staffing*: Recruitment for the VDH-contracted Genetic Counselor position, which was vacated by Allison Schreiber 9/13/2007, is in process. Recruitment for the Virginia Early Hearing Detection and Intervention Program Manager position, which was vacated by Pat Dewey 7/1/2007, is in process. The Birth Defects Prevention and Coordinator position, which was to be established within the Division of Women's and Infants' Health, will not be established. Instead, the duties, which include neural tube defect and fetal alcohol spectrum disorders prevention, will be incorporated into the VCU-contracted Genetic Counselor position.

b. **VDH Virginia Newborn Screening Services (VNSS): A. Greene**
   - *Staffing:* Lillie Chandler, RN, Newborn Screening Nurse, was recently hired. There remains one vacant Newborn Screening Nurse position.
   - *2007 VNSS Data:* From 1/1/2007 through 10/15/2007, the total number of diagnosed cases is 73 (excludes hemoglobinapathies). These include Biotinidase – 2, Cystic Fibrosis - 10, PKU - 3, Galactosemia - 7, Congenital Adrenal Hyperplasia - 4, Homocystinuria – 1, Hyperphenylalninemia - 1, Hypothyroidism - 26, MCADD - 10, Propionic Acid - 1, Partial Biotinidase Deficiency - 5, Profound Biotinidase - 1, and VLCAD - 2.
   - *2007 CCC Referral Data:* From 1/1/2007 through 10/15/2007, the total number of diagnosed cases referred by VNSS to the Care Connection for Children network is 14. These include Blue Ridge CCC - 2, Central Va. CCC – 2, Northern Va. CCC - 9, and Southwest Va. CCC - 1.

c. **VDH: Virginia Sickle Cell Awareness Program: J. Radcliff-Shipman**
   - *Funding:* Over 1,100 children are being followed in the State-funded pediatric comprehensive sickle cell clinics. This represents an increase of 163% in the number of children being followed since the first allocation of $250,000 in 1994. The 2007 General Assembly allocated an additional $200,000 in support of enhanced services to families living with sickle cell disease; however, due to the budget shortfall, VDH proposed a 50% reduction thereby reducing the new funding to $100,000. These cuts will be reflected in a loss of transition services for young people leaving the pediatric program and entering adult care, parent and community education programs and supportive services.
   - *2007 Hemoglobinopathy Data:* As of 10/16/2007, 1,883 trait notification letters have been sent directly to parents of newborns identified with sickle cell trait and hemoglobins C and D trait. The number of parents who have phoned requesting additional information is 11. The number of newborns who have been identified with sickle cell disease (FS,FSA,FSC) and are being tracked for entry into care is 38.

d. **VDH: Children with Special Health Care Needs Program: N. Bullock**
   - *No report.*

e. **Division of Consolidated Laboratory Services: J. Pearson, W. Andrews**
   - *GW-APHL Seminar:* J. Pearson is participating as an instructor at the first event of the George Washington University-American Public Health Laboratories International Institute for Public Health Laboratory Management, a two-week-long seminar that combines teaching

and technical expertise to provide training for senior international public health laboratory leaders.
- *Article:* The article "The Search for the Most Extensive Newborn Screening Panel: What Complexities Will Labs Encounter as Testing Expands?" Clinical Laboratory News, October 2007, Vol. 33, No. 10, was distributed.

f. **March of Dimes: S. Long**
- *No report.*

g. **Part C: M. A. Discenza**
- *No report.*

h. **Virginia Department of Education:** M. C. Ikenberry for T. Campbell
- *Speakers:* Experts in genetics have been conducting presentations to various school nurse educational functions.
- *Standards of Learning:* SOLs may impact how genetics education can be infused in public schools.

6. **Updates: Virginia Genetic and Metabolic Centers**

a. **EVMS: G. Proud**
- *Newborn Screening:* Referrals have increased from about 400 to about 800 per year, mostly due to expanded newborn screening panel.
- *Staffing:* A part-time clinician resigned recently; recruitment is in process. The hospital will support new nurse practitioner and genetic counselor positions.
- *BioMarin:* EVMS is certified to participate in the BioMarin expanded access program for the product sapropterin (a synthetic form of biopterin), an investigational small-molecule oral therapeutic for the treatment of phenylketonuria (PKU).

b. **UVA: B. Wilson, S. Braddock**
- *Newborn Screening:* Like EVMS, referrals continue to increase.
- *Staffing:* New genetic counselor.
- *BioMarin:* Like EVMS, UVA is certified to participate in the BioMarin expanded access program for the product sapropterin.
- *Teratology Information Services (TIS):* A Teratology Information Service is needed in Virginia for healthcare providers. This recommendation was submitted as a public comment for the VDH Title V Block Grant application. N. Ford reported that this will be included in the draft state genetics plan for public health under the Proposed Future Initiatives section. Note: According to the Organization of Teratology Information Services, TIS are comprehensive and multidisciplinary resources for medical consultation on prenatal exposures. TIS interpret information regarding known and potential reproductive risks into risk assessments that are communicated to individuals of reproductive age and health care providers. Source: http://otispregnancy.org/.

c. **VCU: A. Pandya, J. Bodurtha**
- *Newborn Screening:* Like EVMS and UVA, referrals continue to increase. Continue to provide outreach clinics four times a year.
- *Staffing:* W. Nance retired but is still working in the laboratory. Peter O'Connell, Ph.D., Chair, VCU Department of Human Genetics, is stepping down as chair, replacement will likely be another person with a Ph.D.

7. **Next Meeting:**
a. Date: Tuesday, April 1, 2008
b. Time: 10 a.m. – 12 noon
c. Location: Division of Consolidated Laboratories

8. **Adjourn**
Meeting adjourned at 12:10 p.m.